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Case report

A rare case of head injury associated with Albers Schonberg disease

Vipul Namdeorao Ambade MD, LL.B (Associate Professor) [a,*],
Ajit P. Malani MBBS (Senior Resident) [a],
Hemant G. Kukde MBBS (Senior Resident) [a],
Rajkumar N. Meshram MD (Assistant Professor) [b]

[a] *Vasantrao Naik Government Medical College, Department of Forensic Medicine and Toxicology, Yeotmal, Maharashtra State 445 001, India*
[b] *Vasantrao Naik Government Medical College, Yeotmal, Department of Paediatrics and Neonatology, Yeotmal, Maharashtra State 445 001, India*

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Abstract

Albers Schonberg disease commonly called as osteopetrosis is a disorder involving defective bone resorption. The severe autosomal recessive form of osteopetrosis is very rare and characterised by abnormalities in skeletal bones, delayed psychomotor development, optic and facial nerve dysfunction, hydrocephalus, anaemia, haematologic and bleeding disorder, and a diversity of other manifestation. We report a case of intracranial subdural haematoma following a trivial head injury in an infant with severe autosomal recessive form of osteopetrosis.

Keywords: Osteopetrosis; Albers Schonberg disease; Head injury

1. Introduction

Albers-Schonberg first reported osteopetrosis in 1904.[1] It is rare and heterogenous group of disorder characterised by generalised bone sclerosis with thickening and increased fragility of cortical and spongy bone.[2] Two distinct forms of osteopetrosis have been delineated: a severe, autosomal recessive form (an infantile malignant) and a mild, autosomal dominant form (an adult benign). Disturbances of osteoclast function due to mutations in a gene encoding an osteoclast -specific subunit of the vacuolar proton pump *(TCIRG1)* are found in many patients with the recessive form. The dominant form of osteopetrosis has been genetically mapped to chromosome Ip21.[3] In present case, there was intracranial haemorrhage following a trivial head injury in a patient of osteopetrosis.

2. Case report

Six-month boy was brought to private clinic for the first time in June 2004, with complaint of low-grade fever since three weeks and distension of abdomen since neonatal period. At that time, head circumference was 43 cm, weight 5750 g and length 59 cm. There was no history of fits, bleeding tendencies, jaundice, and was excessively irritable. He was the fourth child in the family having previous three normal sisters; all were full term normal deliveries. Developmental milestones were normal till two months. There was a history of second-degree consanguineous marriage of the parent. Pulse, respiration, temperature and blood pressure were normal with mild pallor. There was no icterus, lymphadenopathy, oedema or bleeding spots. RS and CVS examination was within normal limits. Eyes did not fixate. Nystagmus and roving eye movement was present. Pupils were reacting to light.

Blood investigation revealed normocytic erythrocytes with moderate hypochromia and moderate anisopiokilocy-

* Corresponding author. Tel.: +91 9422159573.
E-mail address: vipulambade@rediffmail.com (V.N. Ambade).

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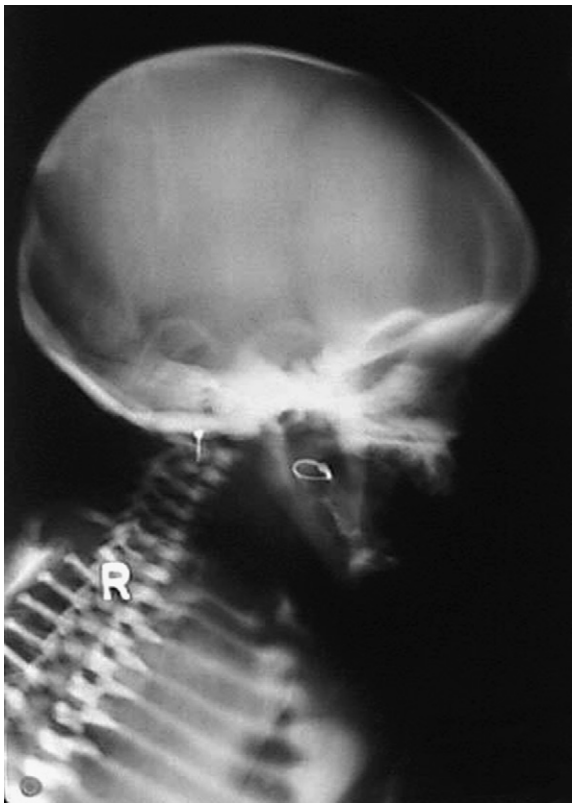

Fig. 1. Radiograph of lateral view of skull showing generalised thickening of base of skull clivus and hard palate.

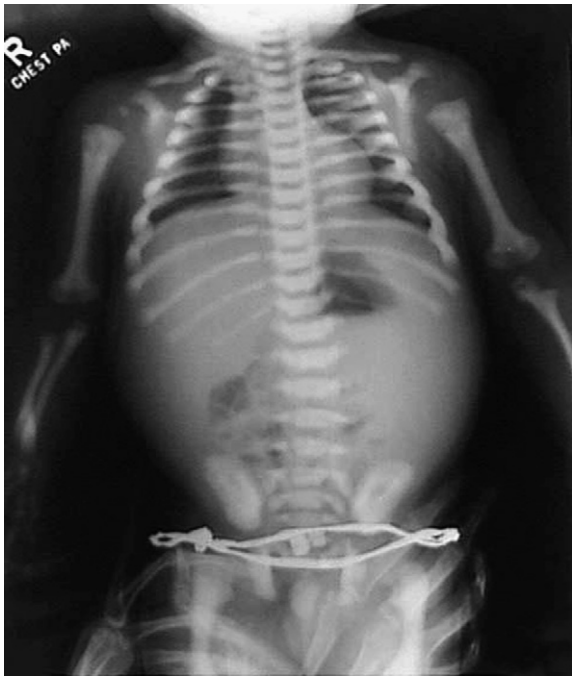

Fig. 2. Radiograph of PA view of chest and abdomen showing generalised increased density with bone within bone appearance and ''rugger jersey'' appearance of spine.

tosis and lymphocytosis with platelets depleted. No parasite seen. ESR (Wintrobe) – 36 mm at end of first hour, Hb – 6.9 g/dl, Platelets – 67×10^9/L, Bleeding time – 8 min, SGPT level – 162 IU/L, Alk. Phosphatase – 1343 IU/L, Serum globulin – 4.4 g/100 ml.

USG abdomen and pelvis showed impression of splenomegaly with no other demonstrable abnormality detected. Radiographs of skull (Fig. 1), chest and abdomen (Fig. 2) showed thickening of base of skull clivus and hard palate with generalised increase density with ''bone within bone'' appearance. Spine showed ''rugger jersey'' appearance and ends of ribs were thickened. Radiograph of right wrist showed linear undisplaced fracture at lower metaphysis of right radius and ulna. A diagnosis of ''osteopetrosis congenita'' – a severe autosomal recessive variety with optic nerve involvement was made. He was started Cap Bio D3 twice daily for three months and was asked to review after three months.

The infant aged about one year was again brought to the clinic in an unconscious condition with history of fall from a height of about one and a half feet from cradle on earthen non-tile surface frequently coated by cow dung. The baby cried after the fall followed by convulsion lasted for 10 min. The mother being nearby promptly attended the baby but within half an hour the infant becomes unconscious. Due to critical condition, he was referred directly to the district hospital as a k/c/o osteopetrosis. On hospitalisation, general condition was poor and there was second episode of generalised tonic clonic convulsion, which was subsided with injection calmpose. Heart rate was 130/m and respiratory rate was 68/m. Pallor and macrocephaly was present with no evidence of external injuries and haemorrhagic tendencies. Pupils were unequal, not responding to light. Deep tendon reflexes were absent. Bilateral crepts were present and liver and spleen were palpable. He was kept on ventilator and was given IV fluids, injection dilantin, crystalline penicillin and gentamycin. But the condition worsened and died after two hours of admission. Body was sent to mortuary for autopsy.

2.1. Autopsy findings

On external examination, abdomen was distended. Teeth were not erupted. Thin built with bony prominences

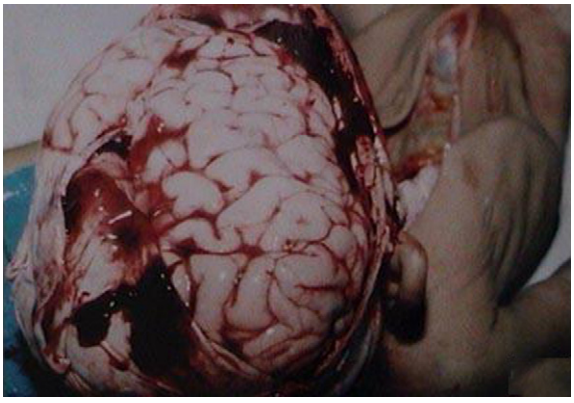

Fig. 3. Photograph of brain on opening of skull vault showing subdural haematoma over right frontal region.

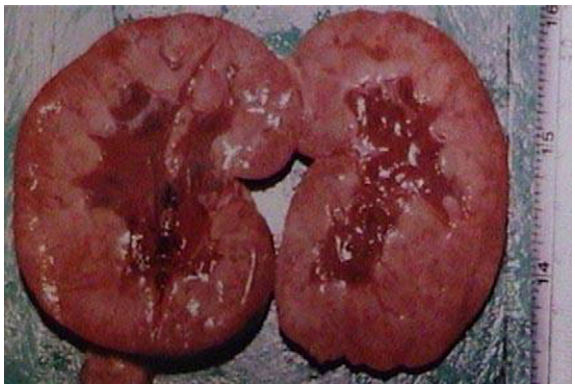

Fig. 4. Photograph of kidneys showing haemorrhagic medulla.

present. Skin was pale and dried, and there was no petechiae or ecchymosis. Post-mortem lividity was not evident. Intravenous injection mark at right cubital fossa with swelling at right wrist. Length was 65 cm, weight ~6 kg, head circumference was 46 cm. There was depressed nasal bridge, prominent malar eminences and widely displaced eyes. On internal examination, red brown haematoma of size 6×4 cm was present under the scalp at right frontal region. Subdural haematoma of about 100 g was present in right frontal region with corresponding flattening of the brain (Fig. 3). Brain was oedematous. There was no fracture in skull bone but showed widespread thickening of base of skull with narrowing of foramina. Lungs showed evidence of bronchopneumonia. Hepato-splenomegaly was present. Visceral organs were pale. There was no evidence of submucosal haemorrhage in the gastrointestinal tract. Para-intestinal lymph nodes were enlarged. Medulla of both kidneys was pulpy and haemorrhagic (Fig. 4). On histopathological examination, kidney shows focal chronic inflammatory infiltrate, liver shows mild periportal chronic inflammatory infiltrate; and bony tissue decalcified.

3. Discussion

This is an unusual case of intracranial haematoma complicating a minor head injury in a patient with known defective bone resorption disorder. Whilst it is recognised that osteopetrosis is associated with anaemia, and hepato-splenomegaly, a heightened risk for subdural haematoma after trivial head injury in a case of osteopetrosis has not been previously described.

Osteopetrosis occurs when abnormal chondroid tissue persists and has a high uptake of calcium. Intestinal absorption of calcium is increased.[4] The severe form of osteopetrosis is usually detected in infancy or earlier and is characterised by macrocephaly, adenoid facies, depressed nasal bridge, hypertelorism, nystagmus, squint, hypotonia, and mental retardation. Milestones are delayed. Bleeding from various sites is also common.[1] There may be generalised lymphadenopathy, hepatosplenomegaly and progressive hydrocephalus may occur. The bone foramina carrying the cranial nerves fail to enlarge with growth so that progressive compression and destruction of nerves occur. Thus, loss of vision, deafness, facial and other paralyses may develop.[5] Radiographs reveal diffuse bone sclerosis. Later films show a bone within bone appearance, which is characteristic. The skull base is thickened and the foramina are narrowed.[3] It involves all skeletal bones and result in haematologic and bleeding disorder and frequent fractures. Radiographs are diagnostic. Neuroimaging often reveals delayed myelination and cerebral atrophy.[2]

The average normal growth parameters of Indian boys at age six-month are weight about 7.1 kg, length 65 cm, head circumference 42.6 cm; and at age 12-month are 8.9 kg weight, 73 cm length, and 45 cm head circumference.[6] Thus there was failure to thrive and macrocephaly in the present case of osteopetrosis. A syndrome of osteopetrosis, renal tubular acidosis, and cerebral calcification is inherited as an autosomal recessive trait. The primary defect in this entity appears to be one of carbonic anhydrase II, one of two enzymes catalysing the association of water and carbondioxide to form bicarbonate.[2] The anaemia in osteopetrosis results from physical loss of marrow space, although this explanation may be too simplistic. This is associated with compensatory extramedullary erythropoiesis in the liver and spleen.[4] Hepatosplenomegaly is a result of myeloid metaplasia, severe anaemia and thrombocytopenia.[1]

Reduction in platelet number constitutes an important cause of generalised bleeding. However spontaneous bleeding does not become evident until the count falls below 20×10^9/L. The common sites of such haemorrhage are the skin and the mucous membranes of the gastrointestinal and genitourinary tracts. Platelet count in the range of 20–50×10^9/L can aggravate post-traumatic bleeding.[7] But in the present case, the platelet was depleted to a level of 67×10^9/L and there were no bleeding spots. Thus it is probable that the individual with osteopetrosis have anaemia and intracranial haemorrhage as a consequence of haematological and bleeding disorder rather than a reflection of any abnormalities in the walls of the blood vessels. As there was platelet deficiency, it is likely to have been responsible for the formation of subdural haematoma in a case of osteopetrosis following a relatively trivial trauma.

Thus it is advocated that even after trivial trauma to head in a patient with known defective bone resorption disorder, intracranial haemorrhage should be suspected due to heightened risk of intracranial bleeding, and care should be taken in that direction at the earliest and expert advice should be sought.

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